MISSOURI
NEWBORN SCREENING

2013 Annual Report

Missouri Department of Health and Senior Services

Governor Jay Nixon

Acknowledgments

The Missouri State Genetic Advisory Committee and its ancillary Newborn Screening Standing Committee, Sickle Cell Standing Committee, Cystic Fibrosis Standing Committee, Newborn Hearing Screening Standing Committee, and the Lysosomal Storage Disorder Task Force Committee play a vital role in supporting the activities of the Missouri Department of Health and Senior Services Newborn Screening Program.

The expertise the committees provide is complemented by department staff who are dedicated to helping Missouri children receive the best care available when diagnosed with one of the serious medical conditions detectable through screening tests.

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Missouri Department of Health and Senior Services
Division of Community and Public Health
Section for Healthy Families and Youth
Bureau of Genetics and Healthy Childhood
and
Missouri State Public Health Laboratory

Table of Contents

Missouri Newborn Screening Disorders

Newborn screening disorders tested and reported in Missouri are as follows:

- Biotinidase deficiency (BIO)
- Classical galactosemia (GALT)
- Congenital adrenal hyperplasia (CAH)
- Congenital primary hypothyroidism (CH)
- Cystic fibrosis (CF)

- Amino Acid Disorders
  - -Arginemia (ARG, arginase deficiency)
  - -Argininosuccinate acidemia (ASA, argininosuccinase)
  - -Citrullinemia type I (CIT-I, argininosuccinate synthetase)
  - -Citrullinemia type II (CIT-II, citrin deficiency)
  - -Defects of biopterin cofactor biosynthesis (BIOPT-BS)
  - -Defects of biopterin cofactor regeneration (BIOPT-RG)
  - -Homocystinuria (HCY, cystathionine beta synthase)
  - -Hyperphenylalaninemia (H-PHE)
  - -Hypermethioninemia (MET)
  - -Maple syrup urine disease (MSUD, branched-chain ketoacid dehydrogenase)
  - -Phenylketonuria (PKU, phenylalanine hydroxylase)
  - -Tyrosinemia type I (TYR-1, fumarylacetoacetate hydrolase)*
  - -Tyrosinemia type II (TYR-II, tyrosine aminotransferase)
  - -Tyrosinemia type III (TYR-III, hydroxyphenylpyruvate dioxygenase)

- Fatty Acid Disorders
  - -Carnitine acylcarnitine translocase deficiency (CACT)
  - -Carnitine uptake defect (CUD, carnitine transport defect)*
  - -Carnitine palmitoyl transferase deficiency I (CPT-1a)
  - -Carnitine palmitoyl transferase deficiency II (CPT-II)
  - -Dienoyl-CoA reductase deficiency (DE-RED)
  - -Glutaric acidemia type II (GA-II, multiple acyl-CoA dehydrogenase deficiency)
  - -Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
  - -Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
  - -Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
  - -Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
  - -Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
  - -Trifunctional protein deficiency (TFP)
  - -Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

The goal of Missouri's newborn screening program is for every newborn to be screened for certain harmful or potentially fatal disorders that aren't otherwise apparent at birth.

- Lysosomal Storage Disorders
  - -Fabry Disease
  - -Gaucher Disease
  - -Hurler Syndrome
  - -Krabbe Disease
  - -Pompe Disease

- Organic Acid Disorders
  - -2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
  - -2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)
  - -3-Hydroxy 3-methylglutaric aciduria (HMG, 3-Hydrox 3-methylglutaryl-CoA lyase)
  - -3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
  - -3-Methylglutaconic aciduria (3MGA, Type I hydratase deficiency)
  - -Beta ketothiolase (BKT, mitochondrial acetoacetyl-CoA thiolase, short-chain ketoacyl thiolase)
  - -Glutaric acidemia type I (GA-1, glutaryl-CoA dehydrogenase)
  - -Isobutyryl-CoA dehydrogenase deficiency (IBG)
  - -Isovaleric acidemia (IVA, Isovaleryl-CoA dehydrogenase)
  - -Malonic acidemia (MAL, malonyl-CoA decarboxylase)
  - -Methylmalonic acidemia (CBL A,B; vitamin B12 disorders)
  - -Methylmalonic acidemia (CBL C,D)
  - -Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)
  - -Multiple carboxylase deficiency (MCD, holocarboxylase synthetase)
  - -Propionic acidemia (PROP, propionyl-CoA carboxylase)

- Hemoglobinopathies
  - -Sickle cell disease (Hb S/S)
  - -Sickle hemoglobin-C disease (Hb S/C)
  - -Sickle beta zero thalassemia disease
  - -Sickle beta plus thalassemia disease
  - -Sickle hemoglobin-D disease
  - -Sickle hemoglobin-E disease
  - -Sickle hemoglobin-O-Arab disease
  - -Sickle hemoglobin Lepore Boston disease
  - -Sickle HPFH disorder
  - -Sickle “Unidentified”
  - -Hemoglobin-C beta zero thalassemia disease
  - -Hemoglobin-C beta plus thalassemia disease
  - -Hemoglobin-E beta zero thalassemia disease
  - -Hemoglobin-E beta plus thalassemia disease
  - -Hemoglobin-H disease

-Homozygous beta zero thalassemia disease
-Homozygous-C disease
-Homozygous-E disorder
-Double heterozygous beta thalassemia disease

- Other
  -Hearing

* There is a lower probability of detection of this disorder during the immediate newborn period.

The Missouri Newborn Screening (NBS) Laboratory's goal is to identify infants at risk and in need of diagnostic testing for the above disorders. A normal screening result does NOT rule out the possibility of an underlying metabolic/genetic disease.

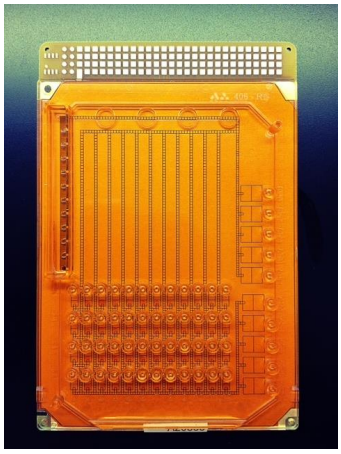
For more details on any of the above-mentioned disorders and how they are screened by the NBS Laboratory, please visit the State NBS Laboratory website at: http://health.mo.gov/lab/newborn.

Screening Spotlight: Missouri's Experience with Full Population Pilot Screening for Pompe Disease, Gaucher Disease, Fabry Disease and Hurler Syndrome using Digital Microfluidics Methodology.

In January 2013, Missouri (MO) began a full population pilot/implementation phase to screen all newborns for four lysosomal storage disorders (LSDs): Pompe disease, Fabry disease, Gaucher disease, and Hurler syndrome. Prior to the onset of screening, an LSD Task Force was formed to assist the Newborn Screening Program with the implementation of adding four LSD's to the newborn screening panel. The LSD Task Force consisted of geneticists, genetic counselors, newborn screening laboratory staff, newborn screening follow-up staff, a chemist, and an adult with Fabry disease.

Missouri elected to use the digital microfluidics (DMF) multiplex enzymatic assay technology provided by Baebies, Inc., formerly Advanced Liquid Logic, Inc. This methodology was selected due to space, cost, and time constraints and the ease at which this methodology could be incorporated into the current newborn screening laboratory. After a full internal review board review and a three month pre-pilot and validation phase, full population pilot screening began on January 11, 2013. All routine newborn screening (NBS) samples received from this date forward were tested for the four LSDs.

For the LSD pilot phase, the MO State Public Health Laboratory required two work stations, each with four DMF instruments. Two scientists were able to fully conduct the LSD testing, interpretation, and reporting. However, four other scientists were cross-trained for backup when necessary.



Cutoffs for flagging abnormal enzyme activities and referrals of positive screens were set conservatively at the beginning of the pilot. Monthly LSD conference calls consisting of the State NBS Laboratory, the NBS Follow-up Program and the genetic tertiary centers were held to provide updates and valuable feedback on the progress of the LSD screening. Cutoffs were adjusted as needed throughout the first year of testing.

Positive screens were referred to one of Missouri's four contracted genetic tertiary centers: Cardinal Glennon Children's Medical Center in St. Louis, Children's Mercy Hospital in Kansas City, St. Louis Children's Hospital in St. Louis, or University Hospitals and Clinics in Columbia. For each referral, the designated center contacted the primary care physician and a plan was then developed to conduct confirmatory testing and any necessary treatment/management for the newborn based on guidelines developed by the MO NBS Program and specialists at the genetic tertiary centers.

On the second day of the pilot testing, a positive Pompe screen was referred to one of the contracted centers and was subsequently confirmed with infantile Pompe through molecular and other diagnostic testing. The newborn was promptly placed on enzyme replacement therapy and is doing well.

From January 11, 2013 to December 31, 2013, over 88,000 newborn samples (approximately 76,000 births) were screened for Pompe disease, Fabry disease, Gaucher disease, and Hurler syndrome in Missouri. A total of 157 newborns were identified with positive screens and were referred to specialists for evaluation and confirmation. Thirty-six newborns were confirmed positive with LSD genotypes: 7 with Pompe (3 infantile and 4 late-onset), 27 with Fabry, 1 with Gaucher, and 1 with Hurler. Eight newborns were confirmed positive with LSD genotypes of unknown significance or genotypes of unknown onset. These 8 newborns were asymptomatic but are continually being followed by the genetic tertiary centers according to established guidelines.

Twenty-four newborns were confirmed positive with pseudodeficiency genotypes, which means that although these newborns display low enzyme levels in the laboratory, they are not affected by the disease in real life and required no further follow-up. Eleven newborns were found to be carriers. A carrier is a person that has inherited only one recessive allele for a genetic condition and does not exhibit symptoms of the disease. Seventy-three newborns were diagnosed as false positives, or normal, based upon normal confirmatory enzyme levels.

However, some of these false positives could have actually been carriers, as several of these newborns displayed low-normal ranges in confirmatory diagnostic enzyme testing and therefore were closed out as normal without further deoxyribonucleic acid (DNA) testing to identify carrier status.

Confirmatory Results from the First Year

Disease	Screened Positive	Confirmed Disorder	Condition Currently of Uknown Significance or Onset	Pseudo-deficiency	Carrier	False Positive*	Lost to Follow-up	Pending
Pompe	33	7 (3 infatile, 4 late)	3	6	8	8	1	0
Gaucher	15	1	2	0	1	11	0	0
Fabry	66	27	3	0	0	33	3	0
Hurler	43	1	0	18	2	21	1	0
Aggregate	157	36	8	24	11	73	5	0

In addition to the screening results and the number of infants identified with LSDs, there were also several other important laboratory findings obtained from this LSD full population pilot phase. The LSD enzyme activities were found to drop slightly during the first 2 weeks of age and then stabilize after 14 days-of-age. Due to this finding, the use of age-related cutoffs for older babies was necessary. Missouri's NBS data has also shown that premature babies can have altered LSD enzyme levels, therefore repeat NBS may be more reliable on this subpopulation.

Multiplexing the four LSD assays was found to be helpful in the assessment of the quality of the NBS sample and the risk for referral, as all four enzyme levels can be observed together. Some seasonal variation was observed with enzyme activities, similar to the galactosemia assay in that more carriers, pseudo-deficiencies, and false positives could be detected during the months where the temperature and the humidity are high. This seasonal phenomenon may have been isolated or sporadic and was not necessarily observed across the full spectrum of NBS samples received during the warmer season.

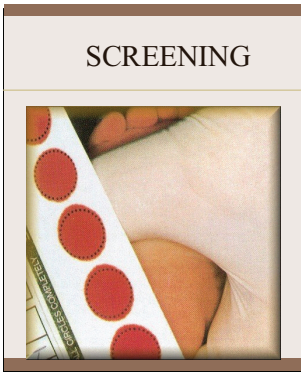
The DMF multiplex method has performed very well for Missouri's screening of approximately 90,000 samples per year, with a positive predictive value averaging 29% and no known missed cases to date. The method was easily incorporated into the Missouri NBS Laboratory and allowed easy cross-training of staff to conduct testing. Excellent communication with the Missouri LSD task force and the genetic tertiary centers promoted ongoing improvements in screening cutoffs and decision schemes.

Missouri has maintained a contract with the New York NBS Program to screen Missouri's samples for Krabbe disease since August 2012. Baebies, Inc. is working to develop fluorimetric assays for Krabbe disease and Niemann-Pick disease for incorporation into the Missouri state LSD testing program.

The Newborn Screening Process

1: TESTING

- The baby's heel is pricked and a few drops of blood are collected on a filter paper 24 to 48 hours after birth.



- The dried blood spot specimen is shipped to the State Public Health Laboratory.
- Specimen is tested for multiple conditions.

2: FOLLOW-UP

- Positive screen results are reported by phone/ fax/letter from lab and follow-up staff to baby's physician. Results are also sent to the appropriate Genetic Tertiary Center in Missouri for follow-up.

- Specimen screening results are entered into data system.
- Baby's physician or health care provider contacts baby's parents.

- Parents bring baby back in for evaluation and more testing at the genetic center.

3: DIAGNOSIS/ INTERVENTION

- Depending on the screen result and the condition screened, repeat or confirmatory testing occurs at the genetic center.

- Parent education for signs/symptoms to watch for is conducted.

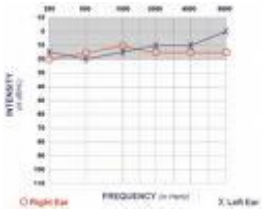
- Baby's physician consults with the specialist appropriate to the condition.

4: TREATMENT & MANAGEMENT

- Once diagnosis is made, treatment begins. For some life-threatening conditions, treatment may occur prior to diagnosis – on the recommendation of a specialist.

- Parents receive treatment guidelines/ education. Team support services as appropriate, include:
  - Metabolic dietitian monitoring and consultation
  - Ongoing blood monitoring
  - Referral to early intervention services
  - Pulmonary/CF services
  - Pediatriac endocrine monitoring
  - Pediatric hematology monitoring
  - Genetic counseling and consideration of family testing
  - Other allied health services as needed

The Newborn Hearing Screening Process

1: SCREENING	2: FOLLOW-UP	3: EVALUATION	4: INTERVENTION
Baby is born. Hospital screens for hearing loss and checks for risk factors for late onset hearing loss prior to discharge.  Hospital submits results to the Missouri Department of Health and Senior Services (DHSS) via the Missouri Electronic Vital Records (MoEVR) system or on a paper form.  DHSS retrieves results from the Missouri Health Strategic Architectures and Information Cooperative (MOHSAIC) data system. 	Hospital reports results to parents and baby's physician.  DHSS sends letters to parents and physicians of newborns who did not pass or who missed the screening.  Parents return baby to hospital/health care provider 1-3 weeks after initial referral. 	Audiologist evaluates babies that don't pass a hearing screening by 3 months of age.  Audiologist reports evaluation results to DHSS.  Audiologist identifies risk factors and makes recommendations.  DHSS sends letter to families of children diagnosed with permanent hearing loss and refers to Missouri's Part C of the Individuals with Disabilities Education Act (IDEA) program, First Steps.	Babies diagnosed with permanent hearing loss enroll in First Steps (early intervention service) by 6 months of age. Babies receive services from the following as appropriate: Primary Care Physician, Otolaryngologist, Geneticist, and Ophthalmologist.  Baby may be a candidate for: hearing aids, cochlear implant, sign language instruction, or speech and language services. 

Newborn Screening Contact Information

Telephone Contacts:

Newborn Screening Laboratory main number	573-751-2662
Order newborn screening specimen forms	573-751-3334
Genetics and Healthy Childhood, for follow-up information	800-877-6246

Web Addresses:

Newborn Screening Laboratory – http://health.mo.gov/lab/newborn

Newborn Blood Spot Screening Program –
http://health.mo.gov/living/families/genetics/newbornscreening

Newborn Hearing Screening Program –
http://health.mo.gov/living/families/genetics/newbornhearing

Appendix 1: Disorders Confirmed for 2013 and Projected Incidence Rates

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Amino Acid Disorders	**12**	**1/6,500***
Arginemia		
Argininosuccinate acidemia	1	
Citrullinemia type I	2	
Citrullinemia type II		
Defects of biopterin cofactor biosynthesis	1	
Defects of biopterin cofactor regeneration		
Homocystinuria	1	
Hypermethioninemia		
Hyperphenylalaninemia		
Hyperphenylalaninemia, benign	3	
Maple syrup urine disease	1	
Maternal PKU		
Phenylketonuria (PKU)	3	
Tyrosinemia type I		
Tyrosinemia type II		
Tyrosinemia type III		
Biotinidase Deficiency (BIOT)	**8**	**1/9,750***
Partial biotinidase deficiency	2	
Profound biotindase deficiency	6	
Congenital adrenal hyperplasia (CAH)	**5**	**1/15,600***
Congenital adrenal hyperplasia non salt water	2	
Congenital adrenal hyperplasia salt water	3	
Congenital primary hypothyroidism (CH)	**49**	**1/1,600**
Cystic fibrosis (CF)	**25**	**1/3,150**
Fatty Acid Oxidation Disorders	**13**	**1/6,000***
Carnitine acylcarnitine translocase deficiency		
Carnitine uptake deficiency		
Carnitine palmitoyl transferase deficiency I		
Carnitine palmitoyl transferase deficiency II		
Dienoyl-CoA reductase deficiency		
Glutaric acidemia type II		
Long-chain hydroxyacyl-CoA dehydrogenase deficiency	1	
Maternal carnitine uptake deficiency		
Medium-chain acyl-CoA dehydrogenase	4	

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
deficiency		
Medium-chain ketoacyl-CoA thiolase deficiency		
Medium/Short chain L-3 hydroxy acyl-CoA dehydrogenase deficiency		
Short-chain acyl-CoA dehydrogenase deficiency	5	
Trifunctional protein deficiency		
Very-long chain acyl-CoA dehydrogenase deficiency	3	
Galactosemia (GALT)	**17**	**1/39,000****
Classical galactosemia	2	
Duarte galactosemia	15	
Lysosomal Storage Disorders (LSD)	**47**	**1/1,700***
Fabry Disease	**30**	
Fabry	26	
Unknown onset	1	
Genotype of unknown significance	3	
Gaucher Disease	**3**	
Gaucher type 1 (non-neuropathic)	1	
Genotype of unknown significance	2	
Hurler Syndrome	**1**	
Hurler Syndrome - severe	1	
Krabbe Disease	**3**	
Genotype of unknown significance	2	
Krabbe unknown risk of onset	1	
Pompe Disease	**10**	
Classical Infantile Onset	2	
Non-classical infantile onset	1	
Later onset	4	
Unknown onset	1	
Genotype of unknown significance	2	
Organic Acid Disorders	**9**	**1/8,700***
2-Methyl-3-hydroxybutyric aciduria		
2-Methylbutyryl-CoA dehydrogenase deficiency		
3-Hydroxy 3-methylglutaric aciduria	1	
3-Methylcrotonyl-CoA carboxylase deficiency		
3-Methylglutaconic aciduria		
Beta ketothiolase		
Glutaric acidemia, type I		
Isobutyryl-CoA dehydrogenase deficiency		

DISORDER	DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE	PROJECTED INCIDENCE RATE
Isovaleric acidemia	1	
Malonic acidemia		
Methylmalonic acidemia (CBL A,B; vitamin B12 disorders)		
Methylmalonic acidemia (CBL, C,D)	2	
Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)		
Multiple carboxylase deficiency		
Propionic acidemia	2	
Forminioglutamic acid (FIGLU) not a disorder on the newborn screening panel but is found	3	
Hemoglobinopathies	**31**	**1/2,500***
Sickle cell anemia disease (Hb S/S)	14	**1/3,000** Total population **1/400** African-American population
Sickle hemoglobin-C disease (FSC)	11	
Sickle beta zero thalassemia disease		
Sickle beta plus thalassemia disease (FSA)	1	
Sickle hemoglobin-D disease		
Sickle hemoglobin-E disease		
Sickle hemoglobin-O-Arab disease		
Sickle hemoglobin Lepore Boston disease		
Sickle HPFH disorder		
Sickle "Unidentified"	2	
Homozygous-C disease (FC)		
Hemoglobin-C beta zero thalassemia disease		
Hemoglobin-C beta plus thalassemia disease	2	
Homozygous-E disorder (FE)		
Hemoglobin-E beta zero thalassemia disease		
Hemoglobin-E beta plus thalassemia disease	1	
Homozygous beta zero thalassemia disease		
Double heterozygous beta thalassemia disease		
Hemoglobin-H disease (Highly Elevated Barts)		
Other (FSX) compound heterozygous Hb S and G-Taipei		

*Combined incidence of all disorders in this category

**Incidence only for classical galactosemia

Appendix 2: Newborn Screening Laboratory Report
Samples Received 2013

	Newborn Samples Received			
	Initial	**Repeat**	**Poor Quality**	**Total Infant Samples**
Jan	6,529	1,161	187	7,877
Feb	5,356	1,048	166	6,570
Mar	5,976	1,230	144	7,350
Apr	6,187	1,266	147	7,600
May	6,194	1,108	106	7,408
Jun	5,806	1,003	70	6,879
Jul	7,078	1,262	99	8,439
Aug	6,611	1,051	107	7,769
Sep	6,506	1,198	120	7,824
Oct	6,962	1,333	149	8,444
Nov	5,485	1,039	139	6,663
Dec	6,737	1,318	196	8,251
Y.T.D.	**75,427 (82.82%)**	**14,017 (15.39%)**	**1,630 (1.79%)**	**91,074**

CALENDAR YEAR 2013
NEWBORN SCREENING LABORATORY REPORT
ABNORMAL RESULTS

Disorder		Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec	Y.T.D.
BIO	Confirmed	1	0	2	0	1	1	0	0	1	0	2	0	**8**
	High Risk	1	0	2	1	3	2	0	0	1	0	2	0	**12**
	Borderline Risk	4	2	1	3	4	8	6	3	4	1	0	0	**36**
CAH	Confirmed	0	0	0	0	1	0	0	0	2	0	0	2	**5**
	High Risk	2	3	3	3	1	2	3	3	6	1	9	12	**48**
	Borderline Risk	42	37	52	56	50	36	45	57	71	79	62	84	**671**
CF	Confirmed	1	0	1	4	3	1	2	4	3	4	2	0	**25**
	Referred	20	17	19	16	17	15	17	21	12	16	18	15	**203**
	Borderline Risk	91	76	70	63	62	60	55	70	72	96	53	53	**821**
CH	Confirmed	4	3	6	3	3	3	6	3	4	4	4	6	**49**
	High Risk	5	5	8	3	3	4	8	6	5	4	6	7	**64**
	Borderline Risk	111	98	158	102	87	93	115	114	119	153	141	143	**1434**
GAL	Confirmed	3	1	2	3	1	2	3	0	1	1	0	0	**17**
	High Risk	3	1	3	3	1	3	7	0	3	1	1	0	**26**
	Borderline Risk	3	2	3	3	4	6	8	3	2	2	0	3	**39**
AA	Confirmed	2	0	1	1	1	0	2	2	0	2	1	0	**12**
	High Risk	0	1	1	1	0	0	1	1	0	3	2	1	**11**
	Moderate Risk	2	1	0	1	2	0	1	1	0	1	0	4	**13**
	Low Risk	74	53	45	48	43	27	32	43	57	59	51	76	**608**
OA	Confirmed	0	0	3	0	2	1	0	0	1	1	1	0	**9**
	High Risk	0	0	2	0	0	1	0	0	0	1	0	1	**5**
	Moderate Risk	2	0	0	1	2	2	1	1	0	2	0	1	**12**
	Low Risk	71	70	111	60	38	31	32	39	33	52	48	54	**639**
FA	Confirmed	3	0	3	1	1	2	1	0	2	0	0	0	**13**
	High Risk	2	1	1	3	1	2	1	0	2	2	0	0	**15**
	Moderate Risk	2	1	3	3	1	3	4	2	2	2	5	5	**33**
	Low Risk	48	49	53	68	54	50	46	51	51	76	51	55	**652**
Hb	Sickle Cell Disease	3	4	1	2	1	2	2	3	2	0	4	4	**28**
	Other Hemoglobinopathies	0	0	0	1	0	1	0	0	0	0	1	0	**3**
	Abnormal Traits	116	113	133	141	128	121	165	150	134	145	105	153	**1604**
LSD	Confirmed	3	2	2	4	10	3	5	5	4	4	0	1	**47**
	High Risk	7	9	9	17	28	22	11	20	15	20	1	8	**167**
	Krabbe Poly Morphs Only	0	0	0	0	2	1	0	3	0	2	1	0	**9**

BIOT = biotinidase deficiency
CAH = congenital adrenal hyperplasia
CF = cystic fibrosis
CH = congenital hypothyroidism
GALT = galactosemia
AA = amino acid
OA = organic acid
FA = fatty acid
Hb = Hemoglobinopathies
LSD = lysosomal storage disorder

Total Confirmed **216**

Appendix 4: Outcome Data – Newborn Screening Samples and Results

- In 2013 there were 75,427 babies tested in the state newborn screening laboratory. There were 91,074 blood spot samples received in the laboratory. Samples received included:

Initial	Repeat	Poor Quality
75,427	14,017	1,630

- In the process of screening newborns for 70 genetic and metabolic conditions, it is the newborn screening laboratory's role to assess the risk of any abnormal screening by evaluating the marker analytes and the levels that were detected. This risk assessment then dictates different levels of action and follow-up protocols. The three categories of risk and the number of test results falling in these categories during 2013 were:

High Risk	Moderate Risk	Low / Borderline Risk
582 (0.77%)	*67 (0.09%)*	*4,900 (6.5%)*

High Risk – Results are immediately phoned and faxed to the physician of record and to the contracted genetic referral centers for consultation and confirmatory testing. Final laboratory reports are mailed to the facility that submitted the specimen and the physician of record.

Moderate Risk – Results are immediately phoned and faxed to the physician of record And the contracted genetic referral centers for consultation and confirmatory testing. Final laboratory reports are mailed to the facility that submitted the specimen and the physician of record.

Low / Borderline Risk – Final laboratory results are mailed to the physician of record and submitting facility with a comment that a repeat newborn screen is necessary.

- Two hundred and twelve (212) confirmed disorders were diagnosed from these abnormal newborn screen results during 2013.

Appendix 5: 2013 Poor Quality Samples

QUANTITY NOT SUFFICIENT: Quantity of blood on filter not sufficient for testing. Possible causes: Removing filter paper before blood has completely filled circle; not allowing an ample size blood drop to form before applying to filter; inadequate heel stick procedure.	89
INCOMPLETE SATURATION: Uneven saturation; blood did not soak through the filter paper. Possible causes: Removing filter paper before blood has completely filled circle or before blood has soaked through to opposite side; improper capillary tube application; allowing filter paper to come in contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood specimen collection.	669
SPECIMEN ABRADED: Filter scratched, torn or abraded. Possible causes: Improper use of capillary tubes. To avoid damaging the filter paper fibers, do not allow the capillary tube to touch the filter paper. Actions such as "coloring in" the circle, repeated dabbing around the circle, or any technique that may scratch, compress, or indent the paper should not be used.	40
LAYERED CLOTTED OR SUPERSATURATED: Possible causes: Touching the same circle on filter paper to blood drop several times; filling circle on both sides of filter paper; application of excess blood; clotted swirl marks from improper capillary application.	645
DILUTED, DISCOLORED OR CONTAMINATED: Possible causes: Squeezing or milking of area surrounding the puncture site; allowing filter paper to come into contact with gloved or ungloved hands, or substances such as alcohol, formula, antiseptic solutions, water, hand lotion, powder, etc., either before or after blood specimen collection; exposing blood spots to direct heat; allowing blood spots to come into contact with tabletop, etc. while drying the sample.	127
OLD SPECIMEN: Specimen greater than 15 days old when received at State Public Health Laboratory.	23
NO BLOOD: Filter submitted without blood.	5
OLD FORM: Sample received on out-of-date form.	2

FILTER AND FORM BARCODES DO NOT MATCH: Bar code on filter does not match bar code on Newborn Screening Form. Collection forms contain barcodes on demographic, hearing and filter portions. The barcodes may not be altered in any way. If incorrect baby is sampled do not remove filter paper and attach to a different demographic portion. If a sampling error occurs the entire form needs to be voided and sample needs to be recollected on a new form. All barcodes must match laboratory copy, submitter copy, newborn hearing screen, and filter.	2
MISSING, INCOMPLETE OR CONFLICTING PATIENT INFORMATION: Missing, incomplete or conflicting demographic information.	4
SERUM RINGS: Serum separated into clear rings around blood spot. Possible causes: Card dried vertically (on side) instead of flat; squeezing excessively around puncture site; allowing filter paper to come in contact with alcohol, hand lotion, etc.	27
BLOOD ON OVERLAY COVER: Overlay cover came in contact with wet blood specimen. Possible causes: Sample is poor quality status because blood soaked from back of filter onto the gold colored backing of the form. The filter circles are designed to hold a specific quantity of blood. If the wet filter is allowed to come into contact with the paper backing of from, blood can be drawn out of filter making the quantative tests performed by the Newborn Screening Laboratory invalid. It is very important that he wet filter paper does not come into contact with any surface until completely dry.	1
Total Poor Quality Samples Received	1,630 (1.79%)

Appendix 6: Newborn Bloodspot Screening Hemoglobinopathy Report 2013

Specimens Received:

Initial:	75,427	(82.7%)
Repeat:	14,017	(15.4%)
Unsatisfactory:	1,630	(1.8%)
Whole Blood:	145	(0.1%)
Total:	**91,219**	

Significant Results = 1,644					
Sickle Cell Disease		**Other Disease Conditions**		**Trait Conditions**	
FS	14	FCA	2	FAS	1038
FSA	1	FSX	2	FSAINC	33
FSC	11	FEA	1	FAC	291
				FCAINC	9
				FAE	34
				FAD	34
				FAX	160
				FASX	2
				FACX	-
				Slightly Elevated Barts	12
				Other Trait condition	-
Total	**26**	**Total**	**5**	**Total**	**1613**

Geographic Follow-up of Significant Disease

Significant Disease Conditions		
St. Louis Area	20	64.5%
Kansas City Area	7	22.6%
Remainder of MO	4	12.9%
Total	**31***	**100%**

*See Appendix 1

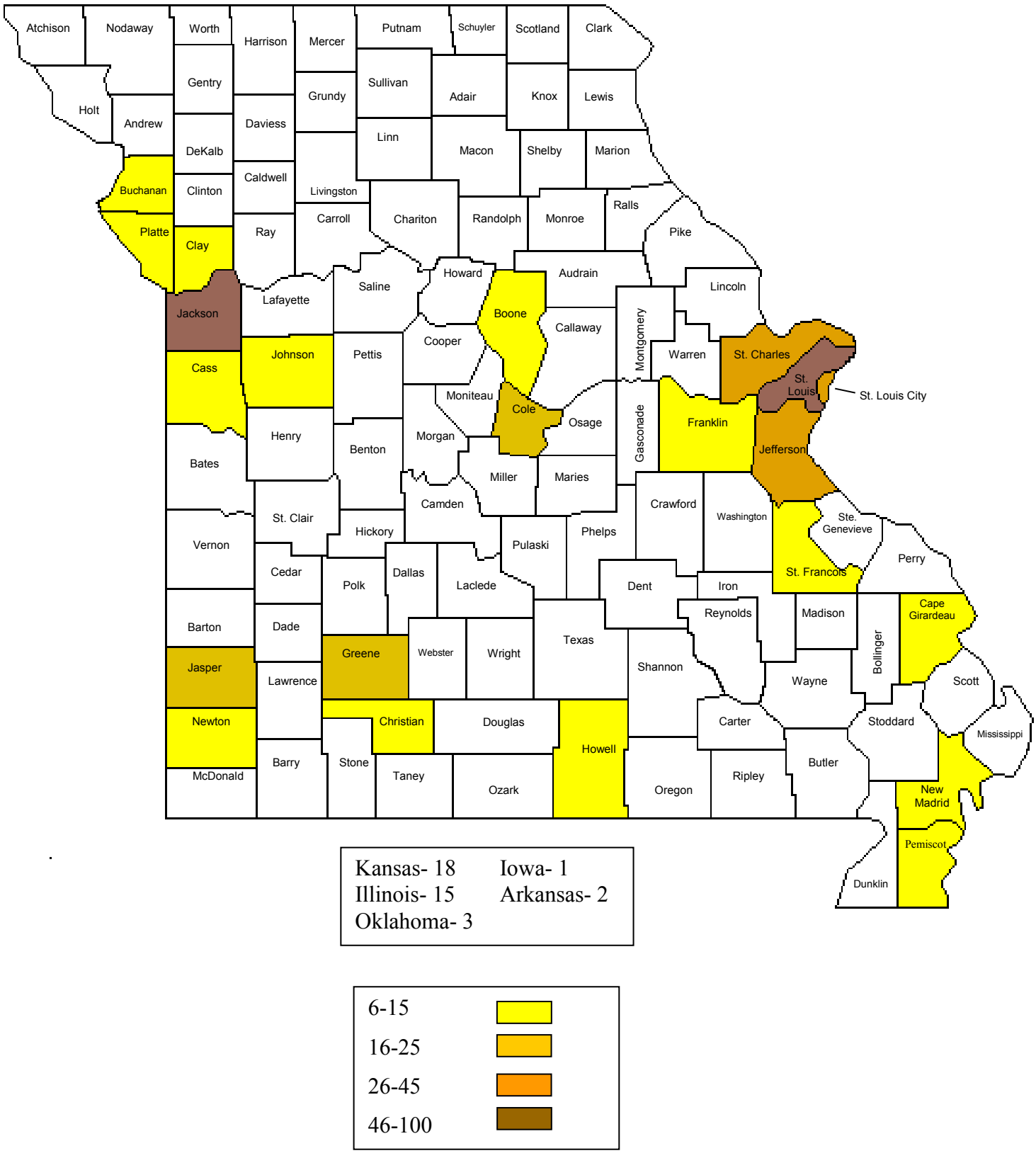
Appendix 7: Missouri Newborn Hearing Screening Data for 2013

2013 calendar year data for Missouri shows:

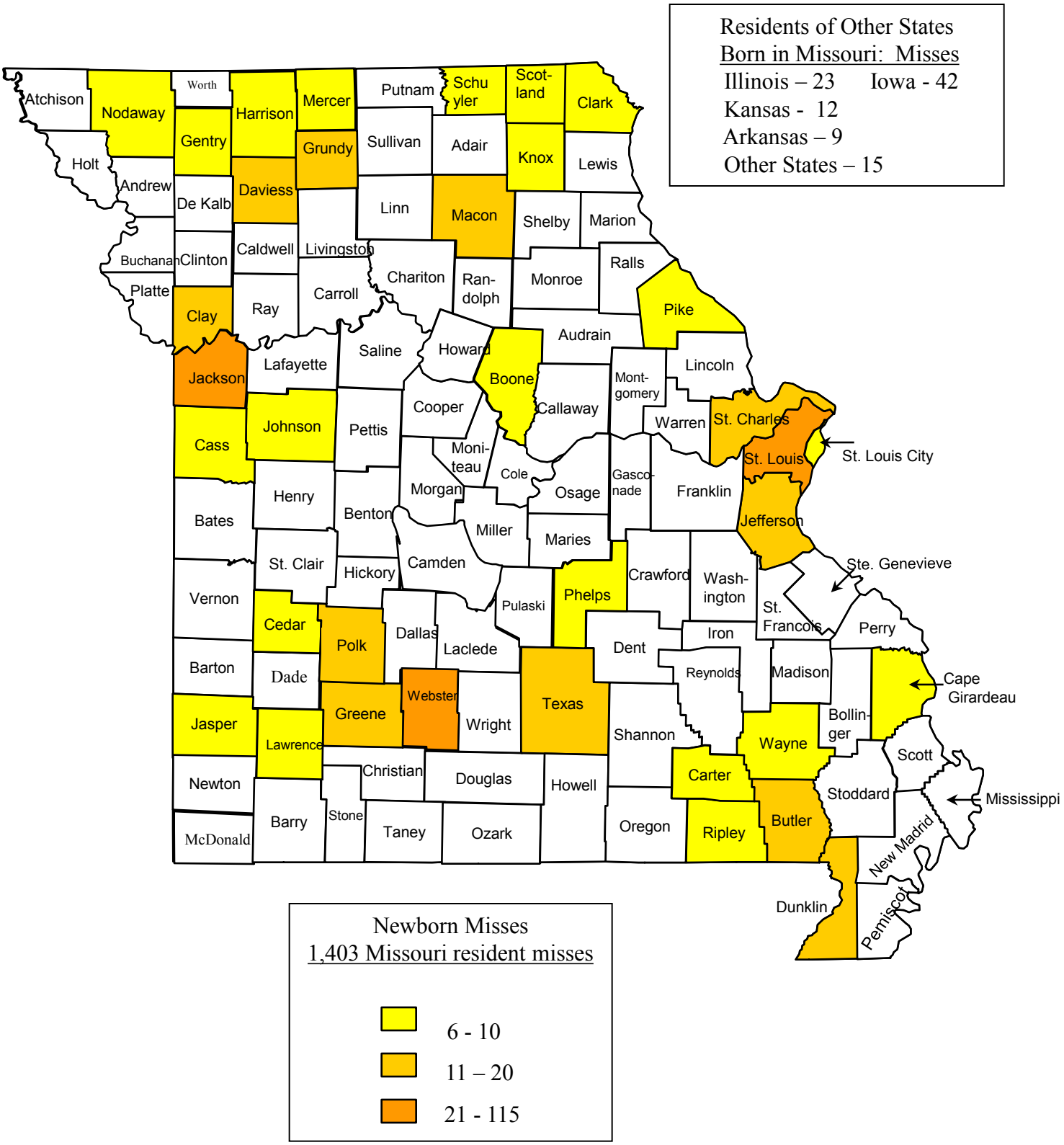
- 76,390 occurrent births (source: Department of Health and Senior Services Vital Records)
- 76,300 occurrent births (source: Missouri Health Strategic Architectures and Information Cooperative [MOHSAIC]*)
- 98.16 percent (74,897) of newborns were screened
- 97.9 percent (73,227) of infants were screened by 1 month of age
- 1.76 percent (1,316) of infants failed the final screening
- 72.97 percent (840) of the infants who failed their final screening and received an audiologic evaluation were evaluated and diagnosed by 3 months of age
- 91 infants were diagnosed with a permanent hearing loss
- 76 infants were enrolled in Missouri's Part C of the Individual with Disabilities Act (IDEA) program, First Steps
- 78.95 percent (60) of the infants enrolled in First Steps did so by 6 months of age

*The difference of 90 births between the occurrent birth count in the program data management system, the Missouri Health Strategic Architectures Information Collaborative (MOHSAIC), and the total occurrent births reported by Vital Records is the result of records that do not yet have an assigned Department Client Number (DCN) and records that are sealed. Records are not released from the Vital Records system to MOHSAIC until the DCN assignment is complete. Non-complete records are due to issues such as paternity and adoptions. Sealed birth records are neither displayed nor counted in MOHSAIC. This report is based upon MOHSAIC records.

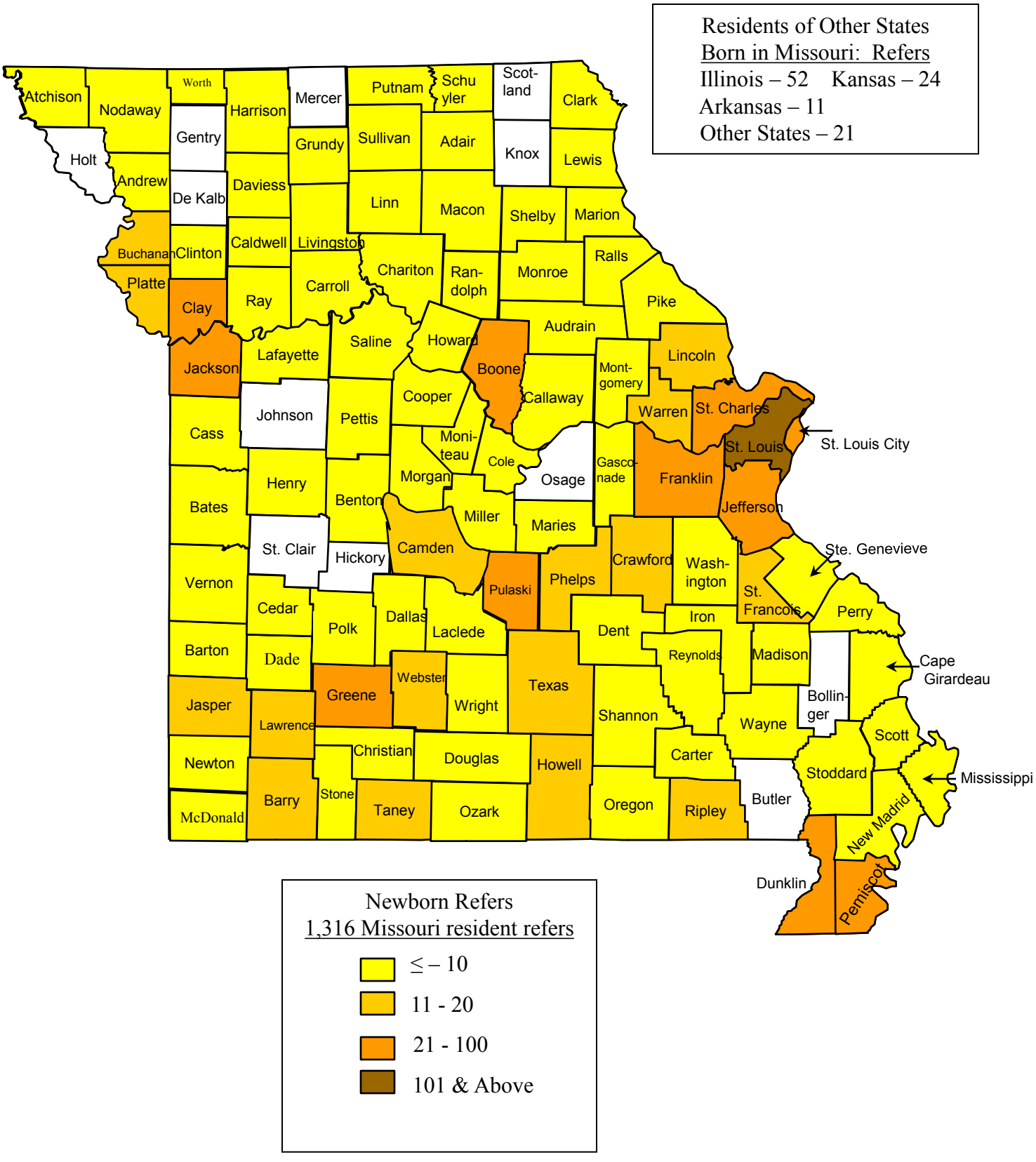
Appendix 8: Number of Newborns with Abnormal Screens Referred for Follow-up by County in 2013



Appendix 9: Number of Newborns that Missed a Hearing Screening by County in 2013



Appendix 10: Number of Newborns Referred after a Hearing Screen by County in 2013



Appendix 11: Newborn Screening Parent Satisfaction Surveys

A satisfaction survey of parents was conducted for families of babies having abnormal newborn screening results reported in 2013. There were 120 satisfaction surveys mailed and 10 were returned for a survey return rate of 8%. Key findings:

Newborn Screening Parent Satisfaction Survey			
	Very Satisfied	Satisfied	Not Satisfied
Staff explained my baby's condition in a way I could understand	89%	11%	
Able to ask questions and discuss decisions about my baby's health care	100%		
Offered reassurance and support	93%	7%	
The treatment staff was knowledgeable	93%	7%	
My questions and concerns were addressed in a timely manner	86%	14%	
The staff provided me with useful referrals and resources	86%	14%	
Received high quality care during my appointments	89%	11%	

A satisfaction survey of parents and children receiving services provided by the hemoglobinopathy resource centers was completed in 2013. There were 1065 surveys mailed and 340 were returned for a survey return rate of 32%. Key findings:

Hemoglobinopathy Resource Center Satisfaction Survey – Parent Response			
	Very Satisfied	Satisfied	Not Satisfied
Treated with respect	97%	1%	2%
Treatment staff was knowledgeable	88%	12%	0%
Questions/concerns addressed in a timely manner	86%	13%	1%
Staff provided useful referrals and resources	83%	15%	2%
Provided with the services needed	97%	2%	1%
Medical care/services received	76%	23%	1%
Received services or treatment without experiencing any problems	97%	0%	3%

Reasons parents responded as not satisfied with services were because of a long wait time. Parents did not indicate what a long wait time meant to them.

Appendix 12: Newborn Hearing Screening Parent Satisfaction Survey

In March 2014* a 2013 satisfaction survey was mailed to parents of children born in Missouri who failed their initial newborn hearing screening between October 2013 and December 2013. There were 578 surveys mailed and 123 were returned for a survey return rate of 21%. The survey examined factors influencing the follow-up time between a failed newborn hearing screening and a repeat screening or an audiologic evaluation.

Key findings:

- 78% of the respondents reported that the birth hospital provided them with written information about the hearing screening prior to the hearing screening.
- 98% of the respondents reported that the birth hospital notified them of the screening result.
- 74% of the respondents reported that the hospital staff explained the importance of knowing whether a baby has a hearing loss early in life.

*Survey conducted every two years.

Missouri Department of Health and Senior Services
P.O. Box 570
Jefferson City, MO 65102
www.health.mo.gov